## Alaska Birth Defect Prevalence Estimates (per 10,000 live births), 2007-2011 birth years (N = 56,218)

**Nationally reportable major congenital anomalies (N=47)**

| Defect | Reports | Report Prevalence (95%CI) | PPV | NPV | Estimated Defects | Defect Prevalence (95%CI)* |
|---|---|---|---|---|---|---|
| Anencephalus | 14 | 2.5 (1.4,4.2) | 0.3929 | 1.0000 | 5.5 | 1.0 (0.4,2.1) |
| Anophthalmia / microphthalmia | 8 | 1.4 (0.6,2.8) | | | | NA |
| Anotia / microtia | 21 | 3.7 (2.3,5.7) | | | | NA |
| Aortic valve stenosis | 8 | 1.4 (0.6,2.8) | | | | NA |
| Atrial septal defect | 888 | 158.0 (147.7,168.7) | | | | NA |
| Atrioventricular septal defect | 41 | 7.3 (5.2,9.9) | | | | NA |
| Biliary atresia | 13 | 2.3 (1.2,4.0) | | | | NA |
| Bladder exstrophy | - | - | | | | NA |
| Choanal atresia | 17 | 3.0 (1.8,4.8) | | | | NA |
| Cleft lip alone | 23 | 4.1 (2.6,6.1) | | | | NA |
| Cleft lip with celft palate | 44 | 7.8 (5.7,10.5) | | | | NA |
| Cleft palate alone | 79 | 14.1 (11.1,17.5) | | | | NA |
| Cloacal exstrophy | - | - | | | | NA |
| Clubfoot | 201 | 35.8 (31.0,41.1) | | | | NA |
| Coarctation of the aorta | 30 | 5.3 (3.6,7.6) | | | | NA |
| Common truncus | 14 | 2.5 (1.4,4.2) | | | | NA |
| Congenital cataract | 29 | 5.2 (3.5,7.4) | | | | NA |
| Congenital posterior urethral valves | 42 | 7.5 (5.4,10.1) | | | | NA |
| Craniosynostosis | - | - | | | | NA |
| Deletion 22 q11 | 5 | 0.9 (0.3,2.1) | | | | NA |
| Diaphragmatic hernia | 32 | 5.7 (3.9,8.0) | | | | NA |
| Double outlet right ventricle | 12 | 2.1 (1.1,3.7) | | | | NA |
| Ebstein anomaly | 7 | 1.2 (0.5,2.6) | | | | NA |
| Encephalocele | 18 | 3.2 (1.9,5.1) | 0.3778 | 1.0000 | 7.1 | 1.3 (0.6,2.6) |
| Esophageal atresia / tracheoesophageal fistula | 15 | 2.7 (1.5,4.4) | | | | NA |
| Gastroschisis | 22 | 3.9 (2.5,5.9) | 0.6174 | 1.0000 | 16.2 | 2.9 (1.8,4.6) |
| Holoprosencephaly | 52 | 9.2 (6.9,12.1) | | | | NA |
| Hypoplastic left heart syndrome | 11 | 2.0 (1.0,3.5) | | | | NA |
| Hypospadias | 327 | 112.6 (100.7,125.5) | 0.8618 | 0.9996 | 291.9 | 100.5 (89.3,112.4) |
| Inerrupted aortic arch | 24 | 4.3 (2.7,6.4) | | | | NA |
| Limb deficiencies | 67 | 11.9 (9.2,15.1) | | | | NA |
| Omphalocele | 78 | 13.9 (11.0,17.3) | 0.1856 | 1.0000 | 14.6 | 2.6 (1.5,4.2) |

| Defect | Reports | Report Prevalence (95%CI) | PPV | NPV | Estimated Defects | Defect Prevalence (95%CI)* |
|---|---|---|---|---|---|---|
| Pulmonary valve atresia and stenosis | 81 | 14.4 (11.4,17.9) | | | | NA |
| Rectal and large intestinal atresia / stenosis | 59 | 10.5 (8.0,13.5) | | | | NA |
| Renal agenesis / hypoplasia | 44 | 7.8 (5.7,10.5) | | | | NA |
| Single ventricle | 8 | 1.4 (0.6,2.8) | | | | NA |
| Small intestinal atresia / stenosis | 42 | 7.5 (5.4,10.1) | | | | NA |
| Spina bifida without anencephalus | 21 | 3.7 (2.3,5.7) | 0.5946 | 0.9999 | 16.0 | 2.8 (1.6,4.4) |
| Teratology of fallot | 34 | 6.0 (4.2,8.5) | | | | NA |
| Total anomalous pulmonary venous sonnection | 10 | 1.8 (0.9,3.3) | | | | NA |
| Transposition of the great arteries | 26 | 4.6 (3.0,6.8) | | | | NA |
| Tricuspid valve atresia and stenosis | 6 | 1.1 (0.4,2.3) | | | | NA |
| Trisomy 13 | 7 | 1.2 (0.5,2.6) | 0.5000 | 1.0000 | 3.5 | 0.6 (0.2,1.6) |
| Trisomy 18 | 12 | 2.1 (1.1,3.7) | 0.5833 | 1.0000 | 7.0 | 1.2 (0.5,2.6) |
| Trisomy 21 (Down syndrome) | 91 | 16.2 (13.0,19.9) | 0.8182 | 0.9999 | 79.9 | 14.2 (11.3,17.5) |
| Turner syndrome | 44 | 7.8 (5.7,10.5) | 0.5217 | 1.0000 | 24.7 | 4.4 (2.9,6.4) |
| Ventricular septal defect | 573 | 101.9 (93.7,110.6) | | | | NA |

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**Ref:** NA = Information not available as of publication; '-'= supressed for cell counts <5; PPV = Positive Predicitve Value; NPV = Negative Predictive value; 95% CI = 95% Confidence Interval

**Note:** The Alaska Birth Defects Registry (ABDR) was established and operates under Alaska statute 7 AAC 27.012. Statue allows reports to by age six years of a child. This report includes defects reported before the 3rd birthday.

**Column descriptions:**
**Defect** = The Nationally reportable birth defect grouping name.
**Reports** = Unless otherwise noted, the number of unique reports by child recieved by ABDR during the specified birth years.
**Report Prevalence (95% CI)** = The number of reported defects divided by the number of Alaska resident in-state births (Children can be reported multiple times. they are counted only once for each condition).
**PPV** = The probability of being defect positive given being reported for the defect.
**NPV** = The probability of being defect negative given not being reported for the defect.
**Estimated Defects** = The estimated number of defects based on the report prevalence, PPV, and 1-NPV.
**Defect Prevalence (95% CI)** = The estimated defect prevalence calculated using a Bayesian approach based on the reported prevalence, PPV and 1-NPV (see formula below).

*Through medical records review and case confirmation of a random sample of reported cases, the defect prevalence is calculated as:

$$PPV(PositivePredictiveValue) = p(defect)|report)$$

$$NPV(NegativePredictiveValue) = p(\overline{defect}|\overline{report})]$$

$$p(defect) \approx [p(report) \cdot PPV] + [p(\overline{report}) \cdot 1 - NPV)]$$

Defect prevalence estimates are a more accurate estimation of the actual diagnosed prevelance of birth defects compared to the reported prevelance estimates in Alaska. ABDR obtains reports from medical providers using International Classification of Disease (ICD) codes that are extracted from individual systems which when aggregated may not reflect true diagnostics. Caution should be used when interpreting and comparing the reported prevalence estimates with national estimates.

**Condition Notes:**
Cleft Lip (CL), Cleft Palate (CP), and Cleft Lip and Palate (CLP) are coded to be mutually exclusive groups.Because an individual can be reported multiple times and for any condition, this coding ensures that for these cleft conditions the classifications conform to the expectation of being mutually exclusive.

- Cleft lip alone includes reports without Cleft palate (CP) or cleft lip and palate (CLP)
- Cleft palate alone includes reports without Cleft lip (CL) or cleft lip and palate (CLP)
- Cleft lip and palate includes reports of both Cleft lip (CL) and Cleft palate (CP), Cleft lip and palate (CLP) or any other combination of Cleft lip (CL) or Cleft palate (CP) with cleft lip and palate (CLP)

Hypospadias is restricted to male births (N = 29,040).

**Resources:** National Birth Defects Prevention Network
Centers for Diesase Control and Prevention

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*Updated: January 12, 2017*
*Code source:* `R:\ABDR\Analysis_New\ABDR_CASECONF\Prevalence07.11.Rmd`